

# TO DETERMINE ETIOLOGY OF GLOBAL DEVELOPMENTAL DELAY IN YOUNG CHILDREN

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## ABSTRACT

**Background:** Developmental delay are group of disorders of early onset estimated to affect 5% to 10% of childhood population. It is commonest neurological condition after ADHD and epilepsy. It describes a clinical presentation that has heterogeneous etiologies and is associated with age specific deficit in learning skills. Accurate etiological determination, despite the fact that many disorders have no specific therapeutic intervention, has specific implementation regarding treatment, prognosis, ongoing medical management of associated conditions, assessment of recurrence risk, counseling of families and implementation of preventive measures. The objective of this study is to determine the common etiologies of global developmental delay so that we can modify our management accordingly.

**Methods:** This study was conducted from January 2014 to March 2015 in Kuwait teaching hospital Peshawar. Patients 6 months to 5 years were enrolled having developmental delay in one or more domain in out patients department. Neurodevelopmental delay was assessed on Denver scal. A detail history and examination carried out. Investigation including CT brain, karyotyping, thyroid function tests, electromyogram, and nerve conduction studies were performed.

**Results:** A total of 170 patients were enrolled in study with developmental delay. Mean age was 32 months (range was 6 months to 5 years). History based evidence shows 90% having developmental delay and 10 % having regression. 101 patients (59.4%) had global developmental delay and 69 children (40.5%) had isolated developmental delay either motor or speech delay. Patient having isolated motor delay were 41 (24.1%) while those having isolated speech delay were 24 (14%). Etiology was identified in 70% of the children, while no specific etiology was identified in 30% of the children. Hypoxic ischemic encephalopathy was the cause of developmental delay in 40 (23.5%) of the children. Brain dysgenesis was present in 17(10%) of the patients. Kernicterus was present in 10 (5.8%) patients. Viral encephalitis and meningitis were present in 7 (4.1%) and 10 (5.8%) respectively. Head trauma was present in 11(6.4%) children. Among the genetic disorder Down syndrome was the commonest cause found in 4(2.3%) children. Neurodegenerative disorder were present in 7(4.1%). 3 (1.8%) children were having TORCH infection. 3 (1.8%) children were having spinal muscular atrophy. Familial motor delay was present among 7 (4.1%) children.

**Conclusion:** Developmental delay need to recognize in children by doing routine check-ups and follow ups at least up to five years. Findings of specific etiologies early would have better outcome in management and recurrence risk assessments.

## INTRODUCTION

Developmental delay are a group of related chronic disorders of early onset estimated to affect 5% to 10% of childhood population<sup>1</sup>. It is far most common cause of referral of a child to pediatric neurologist clinic<sup>2</sup>. Developmental delay is commonest neurological condition after ADHD and epilepsy<sup>3</sup>. About half of children with developmental impairment remain unnoticed before school going. Although severe disorders can be recognized in infancy, it is unusual to diagnose speech impairment, hyperactivity before the age of three or

four years, and learning disabilities are rarely identified before school starts. However an early recognition helps both children and their parents<sup>3</sup>.

Global developmental delay is a subset of developmental disabilities defined as significant delay in two or more of developmental domain that is: motor (gross and fine), speech/language, cognitive, social/personal and activities of daily living<sup>1</sup>. Global developmental delay describes a clinical presentation that has heterogeneous etiologies and is associated with age specific deficits in learning skills<sup>2,3</sup>. The disturbance must be significant that means performance of equal or more below two standard deviation of mean on standardized assessment of development. Incidence is estimated to be 1-3% in under 5 year children<sup>4,5</sup>. Delay in two domains means delay in all domains. When delay is in one domain it is either motor delay or speech impairment. The commonest developmental disorder in children is speech impairment that is about 3-10% of patients<sup>6</sup>. The term global delay is generally reserved for younger children i.e less than 5 years, whereas the term mental retardation is usually applied to older children when

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IQ testing is more valid and reliable. Developmental assessment is an essential component of child care and help us in early diagnosis of a child with global delay and improve outcome<sup>6,7,8</sup>. Children with developmental delay have problems with memory, learning abilities, motor skills, emotional and behaviors.

History in detail and through examination is far most important on identification of a global delay and its etiology leading to establishment of diagnosis in 34% of the patients<sup>2,6</sup>. Further radiological and laboratory investigation may help to define the etiology in rest of the children with delay i-e in 30-40% of the patients<sup>2</sup>. Rest of them may categorized as idiopathic. Accurate etiological determination, despite the fact that many disorders have no specific therapeutic intervention, has specific implementation regarding treatment, prognosis, ongoing medical management of associated conditions, assessment of recurrence risk, counseling of families and implementation of preventive measures<sup>9,10</sup>.

The objective of this study is to determine the common etiologies of global developmental delay so that we can modify our management accordingly.

## MATERIALS & METHODS

This study was conducted from January 2014 to March 2015 in Kuwait teaching hospital Peshawar. Children with developmental delay were identified among children visiting outpatient department of the mentioned hospital. Children aged 6 months to 5 years were enrolled in study having developmental delay either of one or more domains. Neurodevelopmental delay was assessed by Denver scale with comparison to normal peers<sup>5</sup>. The assessment was done by consultant pediatrician to confirm the delay. A detail history with detail clinical examination was carried out by consultant pediatrician for definition of global developmental delay and to find out most probable cause of child condition. Specific laboratory and radiological investigation were performed on a case by case basis, including CT brain, karyotyping, thyroid function tests, EMG, nerve conduction studies. We did not evaluate IQ in these children. Two follow ups was done in a period of 3 months to confirm our diagnosis.

In this study we considered developmental delay in all those children who were having delay as compared to their peers in one or more domains. Cerebral palsy was considered in children having disorder of movement and posture leading to limitation of activity that is due to insult of growing brain (fetal and infant) that is permanent and non-progressive<sup>9</sup>. Cerebral dysgenesis was diagnosed on the CT brain on defining features. Intrapartum and postnatal asphyxia was diagnosed on the basis of comprehensive history and examination and objective documentation of intrapartum complications. Autism is define as spectrum of disorders characterized by difficulties in social interaction, restrictive and repetitive behaviors, no eye contact, difficulty in verbal

and nonverbal communication, insistence of sameness of activity and surroundings, not engage in play with other children, prefer to be alone<sup>6</sup>. These features were identified on history and neurodevelopmental examination. Chromosomal disorders were diagnosed on karyotyping done in the presence of specific features. Developmental speech delay was labeled in children having no other neurological abnormality except speech delay. Those children having history of motor delay in family and have had motor delay without any other abnormality are labeled as familial motor delay.

## RESULTS

A total of 170 patients were enrolled in study with developmental delay. Mean age was 32 months (range was 6 months to 5 years). History based evidence shows 90% having developmental delay and 10 % having regression. 101 patients (59.4%) had global developmental delay and 69 children (40.5%) had isolated developmental delay either moter or speech delay. Patient having isolated moter delay where 41 (24.1%) while those having isolated speech delay were 24 (14.1%).

The parents were relatives in 80% of cases while the history of developmental delay in family was positive in 45% of the patients. The history of seizures was present in 35% of the patients.

Etiology was identified in 70% of the children, while no specific etiology was identified in 30% of the children. Hypoxic ischemic encephalopathy was the cause of developmental delay in 40(23.5%) of the children among them 30(17.6%) were having global delay while 10(5.8%) were having isolated delay. Brain dysgenesis was present in 17(10%) of the patients, among which about 90% were having global delay. Kernicterus was present in 10(5.8%) patient among which half were having global and half were having isolated moter delay. Viral encephalitis and meningitis were present in 7(4.1%) and 10(5.8%) respectively. Head trauma was present in 11(6.4%) of the children. Among the genetic disorder Down syndrome was the commonest found in 4(2.3%) of the children. Neurodegenerative disorder were present in 7(4.1%) of the children, all were having global delay. 3 (1.8%) children were having TORCH infection. 3 (1.8%) children were having spinal muscular atrophy among which 2 were having isolated moter delay and one was having global delay. Famileal moter delay was present among 7 (4.1%) children most of them were having isolated moter delay.

## DISCUSSION

Developmental surveillance is recognized as an integral component of pediatric care. Professional organization dedicated to medical care of the children recommended routine monitoring of the child's developmental progress<sup>4,6</sup>. Parenteral reporting along with the consultant examination constitute the primary means by

**Table 1. The Etiologies of Neurodevelopmental Disability in Studied Groups**

	Global Delay	Isolated Motor Delay	Isolated Speech Delay
Hypoxic Ischemic Encephalopathy	30	10	0
Neurogenerative Disorders	7	0	0
Brain dysgenesis	15	2	0
TORCH	2	1	0
Familial Motor Delay	2	5	0
Spinal Muscular Atrophy	1	2	0
Kernicterus	5	5	0
Genetic Disorders	4	0	0
Viral Encephalitis	7	0	0
Meningitis	10	0	0
Head Trauma	7	2	1
Unknown	29	15	11

which we can identify the children with delay. Regular follow up are than required in high risk and suspicious children. Evidence demonstrate the benefits of early detection of delay and its causes with early intervention if possible to suggest that it may improve outcome. Initial screening is important not only in identifying delay but also differentiate between global delay, speech disorder and autistic spectrum<sup>6,11</sup>.

Accurate etiological determination has specific implication regarding treatment, prognosis, ongoing management of associated condition and recurrence risk assessment, despite the fact that many diseases have no specific therapeutic intervention<sup>12</sup>.

In our study we were able to determine etiology in 2/3 of the cases having developmental delay as same has been reported in previous studies<sup>2,4,6</sup>. Although this percentage could be increased if we had included more specialized and expensive laboratory tests in our study We observe in our study that majority of the patient's were having global delay that is they were delay in two or more than two domain while quite a significant number of children were having isolated delay, among which motor delay was commonest followed by isolated speech delay. Global delay is more frequent as underlying etiology usually effects wide range of areae.g.hypoxia, infection or trauma. Isolated speech delay was present in spectrum of disorders. It is mostly associated with autism. Some were having only maturation delay with rest of intelligence is normal, this is most common in boys. In another study from Iran same findings were observed, autism was observed in quite a number of these patients who were mostly categorized as mentally restarted on GP level<sup>6</sup>. Some of these were also started on antipsychotics drugs.

Etiology of developmental delay has wide spectrum ranging from antenatal insult to postnatal causes.

That's why more than one child has observed in the same family showing genetic predisposition as well. In our study most of the parents were first relative's cousins having history of mental diseases running in families. In this area marriages are mostly arranged among families that is causing increase incidences of mental illnesses in children and adolescence. Hypoxic ischemic encephalopathy, brain dysgenesis, infectious causes and genetic disorders are specific etiologies found in about half of the children. They are easy to diagnose on the history and radiological investigation. A study from India shows chromosomal disorders as the largest group having Down syndrome the commonest one<sup>4</sup>. A study from Canada shows presence of four disorders that is cerebral dysgenesis, hypoxic ischemic encephalopathy, toxin exposure and chromosomal abnormalities for 77% of etiologies in developmental delay<sup>8</sup>. In our study infection and trauma was higher as compared to studies from other areas<sup>4,6,8</sup>. Meningitis and trauma in our setup is quite high and due to lack of expert medical facilities in far flung areas complications are high leading to sequelae. Cerebral palsy was the commonest clinical presentation in our study due to varied underlying causes. These children were having static motor deficit having spastic quadriplegic as the commonest type. A study from India shows cerebral palsy in about 12% of patients categorized in different groups<sup>4</sup>. Majority were having spastic Cerebral palsy. Neurodegenerative disorders are less common in our study as compared to other studies<sup>2,4,8</sup>. TORCH infection was present in quite a few children commonest was being toxoplasmosis. This is same among Asian studies<sup>2,4,6</sup>. A definitive diagnosis was not made 30% of patients in our study. Figures of 10-80 % has been reported in literature<sup>4</sup>. We haven't done specific metabolic screening and genetic techniques in our study as that were quite expensive or not available. A specific

diagnosis is difficult as it needed very expensive or highly specialized techniques. In our study most of the families were from poor socioeconomic group. There might be having high probability of finding a genetic etiology in these undiagnosed children. In our area developmentally delayed children are usually neglected children, one factor might be poverty, where parents are least interested to do advanced investigation or rehabilitation management. Factor of non-acceptance also have important role.

## CONCLUSION

Developmental delay need to recognize in children by doing routine check-ups and follow ups at least up to five years. Findings of specific etiologies early in children have better out come in giving managements and recurrence risk assessments.

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